



SLC12A6 gene

solute carrier family 12 member 6

Normal Function

The *SLC12A6* gene provides instructions for making a protein called a K-Cl cotransporter. This protein is involved in moving charged atoms (ions) of potassium (K) and chlorine (Cl) across the cell membrane. The positively charged potassium ions and negatively charged chlorine ions are moved together (cotransported), so that the charges inside and outside the cell membrane are unchanged (electroneutral).

Electroneutral cotransport of ions across cell membranes is involved in many functions of the body. While the specific function of the K-Cl cotransporter produced from the *SLC12A6* gene is unknown, it seems to be critical for the development and maintenance of nerve tissue. It may be involved in regulating the amounts of potassium, chlorine, or water in cells and intercellular spaces. The K-Cl cotransporter protein may also help regulate the activity of other proteins that are sensitive to ion concentrations.

Health Conditions Related to Genetic Changes

Andermann syndrome

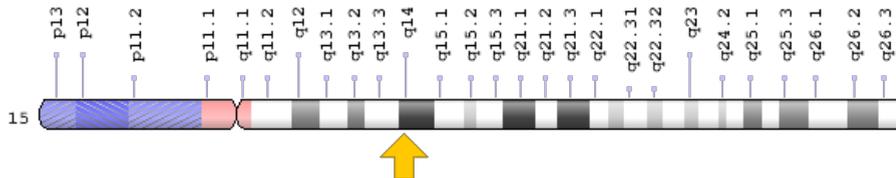
At least six *SLC12A6* gene mutations have been identified in people with Andermann syndrome. Almost all affected individuals of French-Canadian descent have the same mutation in both copies of the *SLC12A6* gene, in which the DNA building block (nucleotide) guanine is deleted at position 2436 (written as 2436delG). This mutation is common in the populations of the Saguenay-Lac-St.-Jean and Charlevoix regions of northeastern Quebec. Most *SLC12A6* gene mutations that cause Andermann syndrome result in a K-Cl cotransporter protein that is shortened and nonfunctional.

The lack of functional protein produced from the *SLC12A6* gene is believed to interfere with the development of the corpus callosum and maintenance of the nerves that transmit signals needed for movement and sensation, resulting in the signs and symptoms of Andermann syndrome.

Chromosomal Location

Cytogenetic Location: 15q14, which is the long (q) arm of chromosome 15 at position 14

Molecular Location: base pairs 34,229,996 to 34,338,064 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACCPN
- agenesis of corpus callosum and peripheral neuropathy (Andermann syndrome)
- DKFZP434D2135
- KCC3
- KCC3A
- KCC3B
- potassium chloride cotransporter 3
- potassium chloride cotransporter KCC3a-S3
- S12A6_HUMAN
- solute carrier family 12 (potassium/chloride transporter), member 6
- solute carrier family 12 (potassium/chloride transporters), member 6
- solute carrier family 12, member 6

Additional Information & Resources

GeneReviews

- Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum
<https://www.ncbi.nlm.nih.gov/books/NBK1372>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC12A6%5BTIAB%5D%29+OR+%28%28KCC3%5BTIAB%5D%29+OR+%28ACCPN%5BTIAB%5D%29+OR+%28KCC3A%5BTIAB%5D%29+OR+%28KCC3B%5BTIAB%5D%29+OR+%28potassium+chloride+cotransporter+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 12 (POTASSIUM/CHLORIDE TRANSPORTER), MEMBER 6
<http://omim.org/entry/604878>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC12A6.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC12A6%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10914
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9990>
- UniProt
<http://www.uniprot.org/uniprot/Q9UHW9>

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